

# Growth Characteristics of Children With Ectodermal Dysplasia Syndromes

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**ABSTRACT.** *Objective.* Clinical observations suggested that growth abnormalities may be present in children with ectodermal dysplasia (ED) syndromes. This study characterizes the longitudinal pattern of growth in a cohort of children with the ED syndromes. We hypothesized that (1) linear and ponderal growth abnormalities are present in children with ED from infancy through adolescence, and (2) linear and ponderal growth abnormalities differ among the clinical variants of these disorders.

*Methods.* We studied 138 children who had ED and were registered with the National Foundation for Ectodermal Dysplasias, 74% of whom had clinical features consistent with the hypohidrotic EDs (HEDs). Height (or length) and weight measurements were obtained by standardized techniques and from review of available medical records. We converted these measurements to weight-for-height (children younger than 5 years and <103 cm in length) or BMI (children  $\geq 2$  years old). Height, weight, weight-for-height, and BMI were converted to age- and gender-specific z scores. We applied linear regression, 1-sample t tests, and analysis of variance to detect linear and ponderal growth abnormalities in children with ED compared with a reference population.

*Results.* Mean weight-for-age, weight-for-height, and BMI-for-age z scores but not height-for-age z score, were significantly lower in children with the ED syndromes than in the reference population. Mean weight-for-age and weight-for-height z scores but not BMI-for-age or height-for-age z scores increased significantly with increasing age. The mean height-for-age z score of children with the ED syndromes other than the HEDs was significantly lower than that of children with the HEDs.

*Conclusions.* Growth abnormalities, measured as weight deficits, were present at an early age in children with the ED syndromes and persisted through adolescence. Height deficits were seen only in children with ED

syndromes other than HEDs. Clinicians should evaluate carefully children with ED syndromes for growth abnormalities. *Pediatrics* 2005;116:e229–e234. URL: [www.pediatrics.org/cgi/doi/10.1542/peds.2004-2830](http://www.pediatrics.org/cgi/doi/10.1542/peds.2004-2830); *height, weight, malnutrition, growth failure, failure to thrive, short stature.*

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ABBREVIATIONS. ED, ectodermal dysplasia; HED, hypohidrotic ectodermal dysplasia; NFED, National Foundation for Ectodermal Dysplasias; EEC, ectrodactyly-ectodermal dysplasia-clefting syndrome; CI, confidence interval.

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The ectodermal dysplasia (ED) syndromes are a group of rare genetic disorders that affect the ectodermal derivatives of the body, including the skin; hair; nails; teeth; and the sebaceous, eccrine, and apocrine glands.<sup>1–4</sup> The hypohidrotic EDs (HEDs), the most common forms of ED, are inherited as X-linked or autosomal recessive disorders, whereas other ED syndromes are inherited as autosomal dominant or recessive disorders.<sup>5,6</sup> The clinical features of the HEDs include sparse, fine hair; missing or conical-shaped teeth; decreased sweat and mucous glands; hypoplastic skin; and heat intolerance with exercise or increased ambient temperature. Treatment is supportive and includes protection from heat exposure; early denture fittings; skin, hair, ear, nose, and nail care; and genetic counseling for family planning.<sup>7,8</sup>

Although many features of the ED syndromes are well known, the pattern of growth in children with ED has not been characterized formally. Growth is the gold standard by which physicians assess the health, development, and well-being of infants and children.<sup>9</sup> A normal growth pattern does not guarantee overall health, but the child with an atypical growth pattern is more likely to present with undesirable complications of the clinical disorder. Clinical observations from the National Foundation for Ectodermal Dysplasias (NFED) suggested that children with ED may have poor linear or ponderal growth.<sup>10</sup> Whether an abnormal growth pattern persists throughout childhood or differs among the clinical variants of the syndrome is unknown.

This research study characterized the longitudinal pattern of growth in a cohort of children with the ED syndromes. We hypothesized that (1) linear and ponderal growth abnormalities are present in children with the ED syndromes from infancy through adolescence and (2) linear and ponderal growth abnor-

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malities differ among the clinical variants of these disorders. We measured the heights and weights of children with ED and supplemented these values with measurements obtained from their available medical records. This report is the first to document weight deficits in early childhood that persist through adolescence in children affected with the ED syndromes and suggests that differences in linear growth may exist among these rare genetic disorders.

## METHODS

### Patients

All children who were from birth to 20 years of age and had the clinical diagnosis of an ED syndrome were eligible for participation. Children were recruited by 1 of 2 procedures. The NFED actively supported the recruitment of children from families who attended its annual family conferences. In addition, the NFED provided the names of children from its membership database for study recruitment. All patients had a diagnosis of ED determined by self-report, review of the medical records by the NFED, and clinical evaluation by physician members of the NFED Scientific Advisory Board. The physicians who confirmed the diagnosis of ED knew each patient and, as specialists in various disciplines, were familiar with the spectrum of manifestations of the ED syndromes. Our sample included 138 children: 72% boys ( $n = 100$ ) and 28% girls ( $n = 38$ ). Most children ( $n = 102$ ; 72%) had a clinical diagnosis of HED (Christ-Siemens-Touraine syndrome).<sup>11</sup> The remainder had 1 of the following diagnoses: ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome ( $n = 6$ ),<sup>12</sup> Clouston syndrome ( $n = 3$ ),<sup>13</sup> Rapp-Hodgkin syndrome ( $n = 4$ ),<sup>14</sup> oculodentodigital syndrome ( $n = 3$ ),<sup>15</sup> hidrotic ectodermal dysplasia ( $n = 1$ ), Hay-Wells syndrome ( $n = 4$ ),<sup>16</sup> keratitis-ichthyosis-deafness syndrome ( $n = 1$ ),<sup>17</sup> Gorlin-Goltz syndrome ( $n = 1$ ),<sup>18</sup> pachyonychia congenita ( $n = 1$ ),<sup>19</sup> or unclassified ( $n = 12$ ).

The institutional review boards for Human Subject Research at Baylor College of Medicine and its affiliated hospitals and at St Louis University each approved the research study. Written informed consent was obtained from the parents or legal guardians of all participant children who were younger than 18 years and directly from all individuals who were older than 18 years.

Individual heights and weights were obtained by direct measurement at the time of examination during the annual family conferences held at several sites in the United States between 2000 and 2003 and indirectly from review of available medical records provided with parental permission by their private physicians. Height measurements, standing, without shoes, were obtained by 2 of us (T.J.F. and T.M.F.) on children who were older than 2 years and were recorded to the nearest 0.1 cm. Length measurements, reclining, without shoes, were obtained by us (T.J.F. and T.M.F.) on children who were younger than 2 years and were recorded to the nearest 0.5 cm. Weight measurements were obtained with an electronic scale, and values were recorded to the nearest 0.1 kg. Heavy clothes and shoes were removed before weighing. BMI was calculated as the ratio of weight divided by height squared ( $\text{kg}/\text{m}^2$ ) for children who were 2 years and older and weight-for-height was calculated for children who were younger than 5 years.<sup>9</sup> Height, weight, weight-for-height, and BMI measurements for all children were converted to age-appropriate  $z$  scores. The method of measurement of values that were obtained from medical records that were provided by private physicians was unknown. The number of measurements for each child varied. Because all data were longitudinal with an irregular interval between each child's measurements, the number of children who contributed data at each age varied. The  $Z$  scores (SD units) for height-for-age, weight-for-age, weight-for-height, and BMI-for-age measurements were calculated from the reference growth standards for the US population established by the National Center for Health Statistics and the Centers for Disease Control and Prevention.<sup>9</sup>

### Data Analysis

Statistical analysis was performed with MiniTab statistical software (Version 13.0; MiniTab Inc, State College, PA) and SPSS

software (Chicago, IL). Height and weight measurements obtained after 20 years of age were excluded from the analysis because these values were beyond the range of the National Center for Health Statistics standards for children. Height and weight data were grouped into 1-month intervals from birth through 20 years. Patients who had multiple observations within a given interval were assigned the average of those observations, thereby allowing each child 1 recording within each interval. The data within each interval were not statistically independent from data in adjacent age intervals and were presented as cross-sectional samples at each interval.

A regression line was fitted through the height-for-age and weight-for-age  $z$  scores for all children from birth to 20 years, weight-for-height  $z$  scores for children who were younger than 5 years, and BMI-for-age  $z$  scores for all children who were aged 2 to 20 years. The formula for these lines was  $y = a + bx$ , where  $y$  indicates height-for-age, weight-for-age, weight-for-height, or BMI-for-age  $z$  score;  $a$  is their respective intercepts;  $b$  is their respective coefficients; and  $x$  is age. These curves were plotted on standard  $z$  score charts for comparison with the reference population. We calculated means, SD, and confidence intervals (CI) of the slopes (presented as change in  $z$  score per month) for height-for-age, weight-for-age, weight-for-height, and BMI-for-age  $z$  scores for all children with ED. We used 1-sample  $t$  tests to detect differences at  $P < .05$  between the mean slopes and the reference mean (0) for the outcome variables.

Subsequently, the incremental change (slope) in height-for-age, weight-for-age, weight-for-height, and BMI-for-age  $z$  scores over time, as measured by age, was calculated by fitting a regression line through the mean height-for-age, weight-for-age, weight-for-height, and BMI-for-age  $z$  scores for each individual who had at least 2 data points per outcome variable. The formula for these lines was  $y = a + bx$ , where  $y$  indicates height-for-age, weight-for-age, weight-for-height, or BMI-for-age  $z$  scores;  $a$  is their respective intercepts;  $b$  is their respective slopes as change in  $z$  score per month; and  $x$  is age. Again, we calculated means, SD, and CIs for changes in height-for-age, weight-for-age, weight-for-height, and BMI-for-age  $z$  scores. We used 1-sample  $t$  tests to detect differences at  $P < .05$  between the mean slopes for individual changes and the reference mean (0) for each of the outcome variables.

We applied 1-way analysis of variance to detect differences ( $P < .05$ ) in the mean height-for-age, weight-for-age, and BMI-for-age  $z$  scores, as well as the mean change (slope) in height-for-age, weight-for-age, and BMI-for-age over time as measured by age, between gender groups and among children who belonged to the different ED syndromes. We used Tukey pairwise comparison tests to detect differences ( $P < .05$ ) between individual ED syndromes with respect to each of the outcome variables.

## RESULTS

The height-for-age, weight-for-age, weight-for-height, and BMI-for-age  $z$  scores of all children with the ED syndromes, plotted for each month of age, and the fitted curves with 95% CIs for each measurement are shown in Figs 1 to 4, respectively. The mean ( $\pm$ SD, 95% CI) height-for-age, weight-for-age, weight-for-height, and BMI-for-age  $z$  scores for all children with the ED syndromes across all ages are summarized in Table 1. The mean weight-for-age, weight-for-height, and BMI-for-age  $z$  scores but not height-for-age  $z$  score of children with the ED syndromes were significantly lower than the reference data for healthy US children. The mean ( $\pm$ SD, 95% CI) height-for-age, weight-for-age, weight-for-height, and BMI-for-age  $z$  scores, calculated as the change (slope) against age in months, for each child individually are summarized in Table 1. The mean change in weight-for-age and weight-for-height  $z$  scores but not height-for-age or BMI-for-age  $z$  scores increased significantly throughout childhood and adolescence.

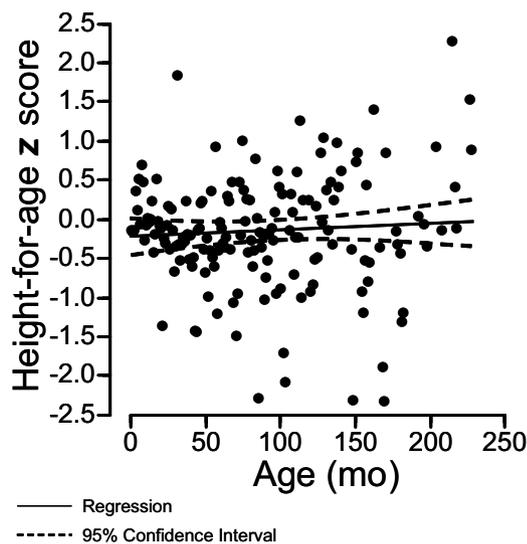


Fig 1. Height-for-age z scores for each month of age in children with the ED syndromes. Height-for-age z score =  $0.229 + 0.001$  age (mo),  $S = 0.730$ ,  $R^2 = 0.4\%$ ,  $R^2(\text{adjusted}) = 0.0\%$ .

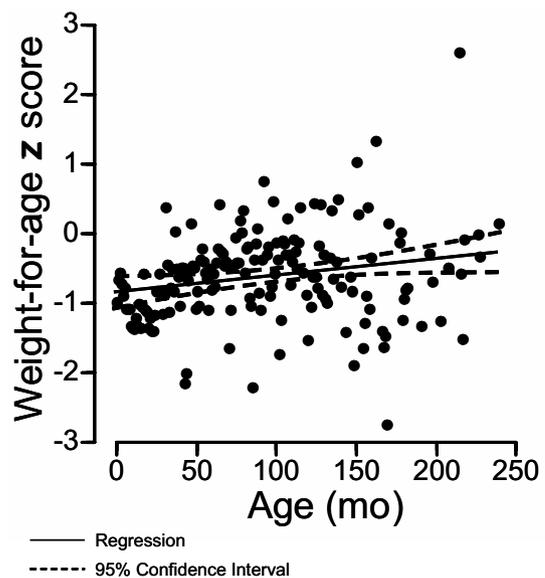


Fig 3. Weight-for-age z scores for each month of age in children with the ED syndromes. Weight-for-age z score =  $-0.839 + 0.002$  age (mo),  $S = 0.648$ ,  $R^2 = 4.1\%$ ,  $R^2(\text{adjusted}) = 3.5\%$ .

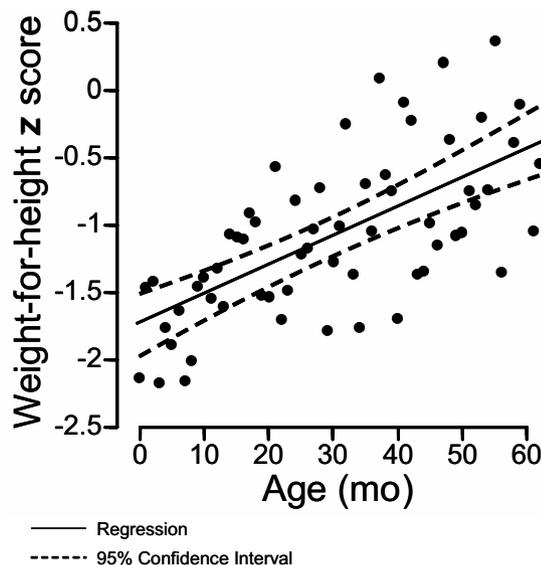


Fig 2. Weight-for-height z scores for each month of age in children with the ED syndromes. Weight-for-height z score =  $-1.757 + 0.023$  age (mo),  $S = 0.453$ ,  $R^2 = 43.9\%$ ,  $R^2(\text{adjusted}) = 43.0\%$ .

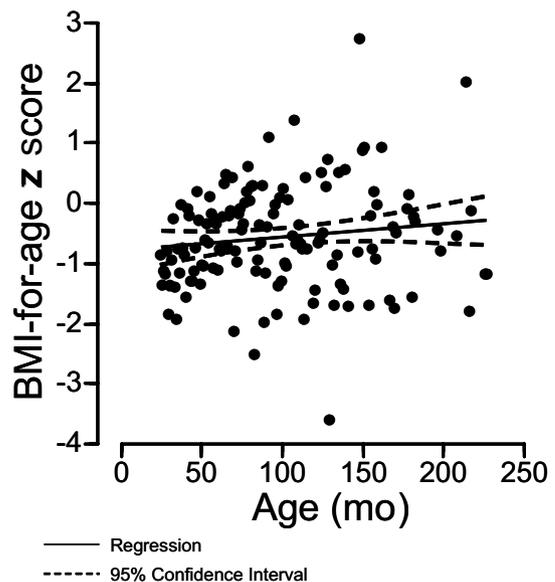


Fig 4. BMI-for-age z scores for each month of age in children with the ED syndromes. BMI-for-age z score =  $-0.779 + 0.002$  age (mo),  $S = 0.867$ ,  $R^2 = 1.6\%$ ,  $R^2(\text{adjusted}) = 0.8\%$ .

The mean and incremental change (slope) in height-for-age, weight-for-age, and BMI-for-age z scores for children with the ED syndromes, based on gender, are summarized in Table 2. The mean and incremental changes in these z scores did not differ significantly between boys and girls who were affected with the ED syndromes. The mean and incremental changes in height-for-age, weight-for-age, and BMI-for-age z scores were examined further for gender differences only in children who were affected with the HEDs. None of these measures differed significantly between boys and girls who were affected with the HEDs (data not shown).

The mean and incremental change (slope) in height-for-age, weight-for-age, and BMI-for-age z scores for children who had a diagnosis of the HED syndromes and all other ED syndromes combined

are summarized in Table 3. The mean and incremental changes in height-for-age z scores but not weight-for-age and BMI-for-age z scores differed significantly between children with the HEDs and all other ED syndromes combined. The mean height-for-age z score was significantly higher but the incremental change in height-for-age z score was significantly lower in children with the HEDs when compared with those with all other ED syndromes combined. The mean height-for-age and weight-for-age z scores for each individual ED syndrome are summarized in Table 4. Height-for-age and weight-for-age comparisons among the individual ED syndrome variants were not performed because of small sample size within these groups.

**TABLE 1.** Mean and Incremental Change Over Time in Height-for-Age, Weight-for-Height, Weight-for-Age, and BMI-for-Age z Scores, Expressed as Group Means, SD, and 95% CIs for Children With the ED Syndromes, Birth to 20 Years, Compared With the Reference Population

Growth Variable	No. of Children	Mean z Score	SD	95% CI	P
Mean (slope)					
Height-for-age	138	-0.119	1.038	-0.294 to 0.055	NS
Weight-for-age	138	-0.655	1.039	-0.830 to -0.481	<.001
Weight-for-height	112	-1.220	0.930	-1.395 to -1.046	<.001
BMI-for-age	124	-0.498	1.060	-0.686 to -0.310	<.001
Incremental change (slope) over time					
Height-for-age	92	0.002	0.033	-0.005 to 0.009	NS
Weight-for-age	94	0.008	0.033	0.002 to 0.015	<.01
Weight-for-height	91	0.380	0.771	0.219 to 0.540	<.001
BMI-for-age	75	0.007	1.060	-0.008 to 0.021	NS

NS indicates not significant.

**TABLE 2.** Mean and Incremental Change Over Time in Height-for-Age, Weight-for-Age, and BMI-for-Age z Scores, Expressed as Group Means and SD: Gender Differences for Children With the ED Syndromes

Growth Variable	No. of Boys	z Score	No. of Girls	z Score	P
Mean (slope)					
Height-for-age	100	-0.07 ± 0.88	38	-0.23 ± 1.38	NS
Weight-for-age	100	-0.65 ± 0.91	38	-0.66 ± 1.33	NS
BMI-for-age	89	-0.51 ± 1.09	35	-0.47 ± 1.00	NS
Incremental change (slope) over time					
Height-for-age	73	0.004 ± 0.035	19	-0.003 ± 0.025	NS
Weight-for-age	75	0.010 ± 0.035	19	0.005 ± 0.029	NS
BMI-for-age	59	0.003 ± 0.069	16	0.020 ± 0.028	NS

NS indicates not significant.

**TABLE 3.** Mean and Incremental Change Over Time in Height-for-Age, Weight-for-Age, and BMI-for-Age z Scores, Expressed as Group Means and SD: Differences Between Children With a Diagnosis of the HED Syndromes and All Other ED Syndromes Combined

Growth Variable	No. of Children With HED Syndromes	z Score	No. of Children With Other ED Syndromes	z Score	P
Mean (slope)					
Height-for-age	102	0.02 ± 0.85	36	-0.52 ± 1.38	<.01
Weight-for-age	102	-0.60 ± 0.88	36	-0.80 ± 1.39	NS
BMI-for-age	96	-0.58 ± 1.05	28	-0.23 ± 1.07	NS
Incremental change (slope) over time					
Height-for-age	80	-0.001 ± 0.027	12	0.025 ± 0.058	<.01
Weight-for-age	82	0.010 ± 0.030	12	-0.003 ± 0.053	NS
BMI-for-age	69	0.007 ± 0.066	6	0.004 ± 0.013	NS

NS indicates not significant.

**TABLE 4.** Mean Height-for-Age and Weight-for-Age z Scores for Individual ED Syndrome Variants

Syndrome	No. of Subjects	Mean Height z Score	Mean Weight z Score
HED	102	0.02 ± 0.85	-0.60 ± 0.88
EEC	6	-0.90 ± 0.84	-1.43 ± 0.58
Clouston	3	0.67 ± 1.02	0.22 ± 1.37
Rapp-Hodgkin	4	-1.66 ± 0.71	-1.55 ± 0.91
Oculodentodigital	3	-1.16 ± 0.79	-2.37 ± 1.74
Hidrotic ectodermal dysplasia	1	-0.10	-1.02
Hay-Wells	4	-1.62 ± 1.58	-1.36 ± 1.69
Keratitits-ichthyosis-deafness	1	-1.60	-1.17
Gorlin-Goltz	1	-0.80	-0.46
Pachyonychia congenita	1	0.01	-0.53
Unclassified	12	0.31 ± 1.50	0.09 ± 1.31

## DISCUSSION

The ED syndromes are a group of rare genetic disorders for which the pattern of growth in children has not been characterized previously. Here we have shown that ponderal growth abnormalities were present at an early age in children with the ED syn-

dromes and persisted through adolescence. Height deficits were seen only in children with the ED syndromes other than the HEDs. Although abnormal linear and ponderal growth patterns have been described in other genetic syndromes,<sup>20-22</sup> reports of growth abnormalities in children with the ED syn-

dromes are infrequent.<sup>23,24</sup> Our findings emphasize to clinicians the importance of monitoring carefully the growth patterns of children with these disorders, although the ED syndromes are uncommon in general pediatric practice.

The growth pattern of the children with the ED syndromes in this study deviated significantly by z score criteria from that of the healthy US reference population. As a group, weight deficits were most pronounced during early childhood, demonstrated not only by the significantly lower mean weight-for-age z score across all age groups but also particularly by the low mean weight-for-height z score that exceeded  $-1$  SD in children who were younger than 5 years. Although these differences were not extreme, the significant increase in the mean incremental change (slope) in weight-for-height and weight-for-age z scores over time suggests that catch-up growth in terms of weight gain was clinically relevant.

The abnormalities in the growth pattern of children with the ED syndromes were apparent well into early adulthood. Weight deficits persisted through adolescence to 20 years of age, demonstrated by the significantly lower mean weight-for-age z score in the children with ED. Although the incremental change in weight-for-age z score increased significantly over time, that the mean BMI-for-age z score remained significantly lower than the reference population and that the incremental change in BMI-for-age z score did not increase significantly over time suggest that weight gains lagged behind height gains through adolescence. We note, however, that our interpretation of these findings may be biased by the relatively fewer data points after 10 years of age contributing a relatively greater weighted mean value to the fitted curve.

Growth abnormalities were neither gender nor syndrome specific, with the exception of stature. Weight deficits were present equally in boys and girls who were affected with the ED syndromes. Although weight deficits were pervasive across all ED syndrome variants, height deficits affected predominantly children who had ED syndromes other than the HEDs. Each of the ED syndromes could not be evaluated individually because of the small numbers of children with each diagnosis. We believe that our data are representative of the pattern of growth in children who are affected with the ED syndromes. However, our findings may be limited by the accuracy of the growth measurements culled from the medical records of our patients.

Causative factors that are responsible for the growth abnormalities in the ED syndromes have not been identified, although they are likely to be manifold. Genetic factors may be implicated in selected ED syndromes when height deficits are the predominant growth abnormality. Genetic defects that lead to a cluster of malformations may predispose some children with the ED syndromes to linear growth stunting.<sup>23,24</sup> Endocrine abnormalities are unlikely to contribute to linear growth abnormalities, although there is a paucity of information about the hormonal status of individuals who are affected with the ED syndromes.<sup>25,26</sup> Growth hormone deficiency has

been reported in 2 children with the EEC syndrome and isolated absent septum pellucidum.<sup>25</sup>

Nutritional factors are more important in the setting of weight abnormalities, particularly weight-for-height and BMI-for-age deficits.<sup>27</sup> Inadequate dietary intakes may be the consequence of feeding difficulties, particularly in young children who have the ED syndromes and missing or deformed teeth.<sup>28</sup> In a previous report from the NFED, feeding problems were identified in 32 of 47 boys with HED, and 19 of those identified had failure to thrive.<sup>10</sup> In our study, the low weight-for-height and BMI-for-age z scores support, in part, the possibility of nutritional inadequacies as a causative factor for the altered growth pattern of children who are affected with the ED syndromes. Although this study was not designed to identify the nutritional origins of the growth abnormalities, weight-for-height deficits may confer a better health outcome particularly on children who are affected with the HED syndromes because the higher surface area per unit of body weight ( $m^2/kg$ ) allows a greater skin surface through which heat can dissipate. This adaptation might explain why the lower BMI-for-age persists into adulthood, although some improvement in BMI-for-age occurs as these children progress through adolescence.

Gastrointestinal disorders such as constipation and gastroesophageal reflux also may reduce dietary intakes because of abdominal pain and vomiting. In the earlier observational report from the NFED, constipation seemed more prevalent in children who were affected with the HED syndromes compared with unaffected individuals and required dietary intervention.<sup>10</sup> In addition, 4 children required gastrostomy button or nasogastric tube feedings because of severe acid reflux and poor weight gain.<sup>10</sup> We believe that clinicians should assess routinely both the dietary intake of all children with the ED syndromes and the ability to feed adequately. In the presence of weight deficits, the clinician should screen for other underlying disorders and simultaneously institute dietary supplementation with energy- and nutrient-dense formulas and foods.

Abnormalities in the components of energy metabolism do not account for weight-for-height deficits in the majority of children with the ED syndromes. The measurement of thermal heat exchange during sleep in children with anhidrotic ED demonstrated that a fall in metabolic heat production, measured by a fall in rectal temperature, represented a state of energy conservation rather than expenditure.<sup>29</sup> Isolated cases of malabsorption as a consequence of exocrine insufficiency<sup>30</sup> or focal intestinal necrosis and partial villous atrophy<sup>31</sup> have been described. Although these latter cases account for poor weight gain in some children who are affected with the ED syndromes, none of the children in our study had diarrhea associated with fecal fat loss.

## CONCLUSIONS

Linear and ponderal growth abnormalities were prevalent in children who were affected with the ED syndromes. As a group, weight deficits were present at an early age and persisted through adolescence.

Height deficits were seen only in children with ED syndromes other than the HEDs. Causal factors that result in these abnormal patterns of growth are unknown. These observations provide novel information about the pattern of growth in children with these rare genetic disorders. Children with ED syndromes warrant careful attention and evaluation for linear and ponderal growth abnormalities by the clinician.

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